chylomicron retention disease

Chylomicron retention disease is an inherited disorder that affects the absorption of dietary fats, cholesterol, and certain fat-soluble vitamins. As food is digested after a meal, molecules called chylomicrons are formed to carry fat and cholesterol from the intestine into the bloodstream. Chylomicrons are also necessary for the absorption of certain fat-soluble vitamins, such as vitamin E and vitamin D. A lack of chylomicron transport causes severely decreased absorption (malabsorption) of dietary fats and fat-soluble vitamins. Sufficient levels of fats, cholesterol, and vitamins are necessary for normal growth and development.

The signs and symptoms of chylomicron retention disease appear in the first few months of life. They can include failure to gain weight and grow at the expected rate (failure to thrive); diarrhea; and fatty, foul-smelling stools (steatorrhea). Other features of this disorder may develop later in childhood and often impair the function of the nervous system. Affected people may eventually develop decreased reflexes (hyporeflexia) and a decreased ability to feel vibrations.

Frequency

Chylomicron retention disease is a rare condition with approximately 40 cases described worldwide.

Genetic Changes

Mutations in the *SAR1B* gene cause chylomicron retention disease. The *SAR1B* gene provides instructions for making a protein that is involved in transporting chylomicrons within enterocytes, which are cells that line the intestine and absorb nutrients.

SAR1B gene mutations impair the release of chylomicrons into the bloodstream. A lack of chylomicrons in the blood prevents dietary fats and fat-soluble vitamins from being used by the body, leading to the nutritional and developmental problems seen in people with chylomicron retention disease.

Inheritance Pattern

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Anderson disease
- Anderson syndrome
- CMRD
- hypobetalipoproteinemia with accumulation of apolipoprotein B-like protein in intestinal cells
- lipid transport defect of intestine

Diagnosis & Management

These resources address the diagnosis or management of chylomicron retention disease:

- Genetic Testing Registry: Chylomicron retention disease
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0795956/
- MedlinePlus Encyclopedia: Malabsorption
  https://medlineplus.gov/ency/article/000299.htm

These resources from MedlinePlus offer information about the diagnosis and management of various health conditions:

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html
- Palliative Care
  https://medlineplus.gov/palliativecare.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Malabsorption
  https://medlineplus.gov/ency/article/000299.htm
- Health Topic: Malabsorption Syndromes
  https://medlineplus.gov/malabsorptionsyndromes.html
- Health Topic: Metabolic Disorders
  https://medlineplus.gov/metabolicdisorders.html

Genetic and Rare Diseases Information Center

- Chylomicron retention disease
  https://rarediseases.info.nih.gov/diseases/9683/chylomicron-retention-disease

Educational Resources

- Colorado State University: Fat-Soluble Vitamins
  http://extension.colostate.edu/topic-areas/nutrition-food-safety-health/fat-soluble-vitamins-a-d-e-and-k-9-315/
- Disease InfoSearch: Chylomicron Retention Disease
  http://www.diseaseinfosearch.org/Chylomicron+Retention+Disease/1633
- MalaCards: chylomicron retention disease
  http://www.malacards.org/card/chylomicron_retention_disease
- Merck Manual Consumer Version: Hypolipidemia
  http://www.merckmanuals.com/home/hormonal-and-metabolic-disorders/cholesterol-disorders/hypolipidemia
- Merck Manual Consumer Version: Overview of Malabsorption
  http://www.merckmanuals.com/home/digestive-disorders/malabsorption/overview-of-malabsorption
- Orphanet: Chylomicron retention disease
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=71

Patient Support and Advocacy Resources

- CLIMB: Children Living with Inherited Metabolic Diseases
  http://www.climb.org.uk/

Genetic Testing Registry

- Chylomicron retention disease
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0795956/

ClinicalTrials.gov

- ClinicalTrials.gov
  https://clinicaltrials.gov/ct2/results?cond=%22chylomicron+retention+disease%22

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28chylomicron+retention+disease%5BTIAB%5D%29+OR+%28cmrd%5BTIAB%5D%29+OR+%28anderson+disease%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D

OMIM

- CHYLOMICRON RETENTION DISEASE
  http://omim.org/entry/246700

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